

SEQUENCE LISTING

<110> DREYFUSS, Gideon
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 PELLIZZONI, Livio
 FISCHER, Utz
 Liu, Qing

<120> COMPOSITIONS, METHODS AND KITS USEFUL FOR THE DIAGNOSIS AND
 TREATMENT OF SPINAL MUSCULAR ATROPHY

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<140> US 09/399,081

<141> 1999-09-17

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 ccttcagatt gcagactgaa gccaggaag atgattggta tgactgtcat agggaaatac 2580
 gtctgagttt ttctgatacc tatcaggatt atgaggagta ctggagagct tactacaggg 2640
 catggcaaga atattatgct gccgcttctc attcatatta ttggaatgct cagagacatc 2700
 caagttggat ggcagcttat cacatgaata ccatttatct acaagaaatg atgcatagta 2760
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<210> 8
 <211> 824
 <212> PRT
 <213> Homo sapiens

<400> 8

Met	Ala	Ala	Ala	Phe	Glu	Ala	Ser	Gly	Ala	Leu	Ala	Ala	Val	Ala	Thr
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Ala	Met	Pro	Ala	Glu	His	Val	Ala	Val	Gln	Val	Pro	Ala	Pro	Glu	Pro
			20					25					30		
Thr	Pro	Gly	Pro	Val	Arg	Ile	Leu	Arg	Thr	Ala	Gln	Asp	Leu	Ser	Ser
		35					40				45				
Pro	Arg	Thr	Arg	Thr	Gly	Asp	Val	Leu	Leu	Ala	Glu	Pro	Ala	Asp	Phe
	50				55					60					
Glu	Ser	Leu	Leu	Leu	Ser	Arg	Pro	Val	Leu	Glu	Gly	Leu	Arg	Ala	Ala
65					70					75				80	

Gly	Phe	Glu	Arg	Pro	Ser	Pro	Val	Gln	Leu	Lys	Ala	Ile	Pro	Leu	Gly	85	90	95
Arg	Cys	Gly	Leu	Asp	Leu	Ile	Val	Gln	Ala	Lys	Ser	Gly	Thr	Gly	Lys	100	105	110
Thr	Cys	Val	Phe	Ser	Thr	Ile	Ala	Leu	Asp	Ser	Leu	Val	Leu	Glu	Asn	115	120	125
Leu	Ser	Thr	Gln	Ile	Leu	Ile	Leu	Ala	Pro	Thr	Arg	Glu	Ile	Ala	Val	130	135	140
Gln	Ile	His	Ser	Val	Ile	Thr	Ala	Ile	Gly	Ile	Lys	Met	Glu	Gly	Leu	145	150	155
Glu	Cys	His	Val	Phe	Ile	Gly	Gly	Thr	Pro	Leu	Ser	Gln	Asp	Lys	Thr	165	170	175
Arg	Leu	Lys	Lys	Cys	His	Ile	Ala	Val	Gly	Ser	Pro	Gly	Arg	Ile	Lys	180	185	190
Gln	Leu	Ile	Glu	Leu	Asp	Tyr	Leu	Asn	Pro	Gly	Ser	Ile	Arg	Leu	Phe	195	200	205
Ile	Leu	Asp	Glu	Ala	Asp	Lys	Leu	Leu	Glu	Glu	Gly	Ser	Phe	Gln	Glu	210	215	220
Gln	Ile	Asn	Trp	Ile	Tyr	Ser	Ser	Leu	Pro	Ala	Ser	Lys	Gln	Met	Leu	225	230	235
Ala	Val	Ser	Ala	Thr	Tyr	Pro	Glu	Phe	Leu	Ala	Asn	Ala	Leu	Thr	Lys	245	250	255
Tyr	Met	Arg	Asp	Pro	Thr	Phe	Val	Arg	Leu	Asn	Ser	Ser	Asp	Pro	Ser	260	265	270
Leu	Ile	Gly	Leu	Lys	Gln	Tyr	Tyr	Lys	Val	Val	Asn	Ser	Tyr	Pro	Leu	275	280	285
Ala	His	Lys	Val	Phe	Glu	Glu	Lys	Thr	Gln	His	Leu	Gln	Glu	Leu	Phe	290	295	300
Ser	Arg	Ile	Pro	Phe	Asn	Gln	Ala	Leu	Val	Phe	Ser	Asn	Leu	His	Ser	305	310	315
Arg	Ala	Gln	His	Leu	Ala	Asp	Ile	Leu	Ser	Ser	Lys	Gly	Phe	Pro	Ala	325	330	335
Glu	Cys	Ile	Ser	Gly	Asn	Met	Asn	Gln	Asn	Gln	Arg	Leu	Asp	Ala	Met	340	345	350
Ala	Lys	Leu	Lys	His	Phe	His	Cys	Arg	Val	Leu	Ile	Ser	Thr	Asp	Leu	355	360	365
Thr	Ser	Arg	Gly	Ile	Asp	Ala	Glu	Lys	Val	Asn	Leu	Val	Val	Asn	Leu	370	375	380

Asp	Val	Pro	Leu	Asp	Trp	Glu	Thr	Tyr	Met	His	Arg	Ile	Gly	Arg	Ala	
385					390					395					400	
Gly	Arg	Phe	Gly	Thr	Leu	Gly	Leu	Thr	Val	Thr	Tyr	Cys	Cys	Arg	Gly	
				405					410						415	
Glu	Glu	Glu	Asn	Met	Met	Met	Arg	Ile	Ala	Gln	Lys	Cys	Asn	Ile	Asn	
			420					425					430			
Leu	Leu	Pro	Leu	Pro	Asp	Pro	Ile	Pro	Ser	Gly	Leu	Met	Glu	Glu	Cys	
		435					440					445				
Val	Asp	Trp	Asp	Val	Glu	Val	Lys	Ala	Ala	Val	His	Thr	Tyr	Gly	Ile	
	450					455					460					
Ala	Ser	Val	Pro	Asn	Gln	Pro	Leu	Lys	Lys	Gln	Ile	Gln	Lys	Ile	Glu	
465					470					475					480	
Arg	Thr	Leu	Gln	Ile	Gln	Lys	Ala	His	Gly	Asp	His	Met	Ala	Ser	Ser	
				485					490						495	
Arg	Asn	Asn	Ser	Val	Ser	Gly	Leu	Ser	Val	Lys	Ser	Lys	Asn	Asn	Thr	
			500					505					510			
Lys	Gln	Lys	Leu	Pro	Val	Lys	Ser	His	Ser	Glu	Cys	Gly	Ile	Ile	Glu	
		515					520					525				
Lys	Ala	Thr	Ser	Pro	Lys	Glu	Leu	Gly	Cys	Asp	Arg	Gln	Ser	Glu	Glu	
	530					535					540					
Gln	Met	Lys	Asn	Ser	Val	Gln	Thr	Pro	Val	Glu	Asn	Ser	Thr	Asn	Ser	
545					550					555					560	
Gln	His	Gln	Val	Lys	Glu	Ala	Leu	Pro	Val	Ser	Leu	Pro	Gln	Ile	Pro	
				565					570					575		
Cys	Leu	Ser	Ser	Phe	Lys	Ile	His	Gln	Pro	Tyr	Thr	Leu	Thr	Phe	Ala	
			580					585					590			
Glu	Leu	Val	Glu	Asp	Tyr	Glu	His	Tyr	Ile	Lys	Glu	Gly	Leu	Glu	Lys	
		595					600					605				
Pro	Val	Glu	Ile	Ile	Arg	His	Tyr	Thr	Gly	Pro	Gly	Asp	Gln	Thr	Val	
	610					615					620					
Asn	Pro	Gln	Asn	Gly	Phe	Val	Arg	Asn	Lys	Val	Ile	Glu	Gln	Lys	Val	
625					630					635					640	
Pro	Val	Leu	Ala	Ser	Ser	Ser	Gln	Ser	Gly	Asp	Ser	Glu	Ser	Asp	Ser	
				645					650					655		
Asp	Ser	Tyr	Ser	Ser	Arg	Thr	Ser	Ser	Gln	Ser	Lys	Gly	Asn	Lys	Ser	
			660					665					670			
Tyr	Leu	Glu	Ser	Ser	Ser	Asp	Asn	Gln	Leu	Lys	Asp	Ser	Glu	Ser	Thr	
		675					680					685				

Pro Val Asp Asp Arg Ile Ser Leu Glu Gln Pro Pro Asn Gly Thr Asp
 690 695 700
 Thr Pro Asn Pro Glu Lys Tyr Gln Glu Ser Pro Gly Ile Gln Met Lys
 705 710 715 720
 Thr Arg Leu Lys Glu Gly Ala Ser Gln Arg Ala Lys Gln Ser Arg Arg
 725 730 735
 Asn Leu Pro Arg Arg Ser Ser Phe Arg Leu Gln Thr Glu Ala Gln Glu
 740 745 750
 Asp Asp Trp Tyr Asp Cys His Arg Glu Ile Arg Leu Ser Phe Ser Asp
 755 760 765
 Thr Tyr Gln Asp Tyr Glu Glu Tyr Trp Arg Ala Tyr Tyr Arg Ala Trp
 770 775 780
 Gln Glu Tyr Tyr Ala Ala Ala Ser His Ser Tyr Tyr Trp Asn Ala Gln
 785 790 795 800
 Arg His Pro Ser Trp Met Ala Ala Tyr His Met Asn Thr Ile Tyr Leu
 805 810 815
 Gln Glu Met Met His Ser Asn Gln
 820

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 <211> 797
 <212> DNA
 <213> Gallus gallus

<400> 9
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 gggacgacac ggccctcatc aaggcgtagc acaaggcggt ggccctcctc aagaatgctt 120
 taaagaacgg ggactgctca gagccttcgg acaaacagga gcagcgggcg ggggtgaaaa 180
 ggaaaaacag caagaagaac aggaacagaa acaagagcaa cgccgtgccg ttgaagcagt 240
 ggaaagttag cgacagctgt aacgctgttt ggtctgagga tggtaatgtc taccctgcaa 300
 ctattgcctc cataaatctg aagaggggta catgcgttgt tacttacacc ggatatggaa 360
 acaaggagga acagaacctg gctgatctac ttctccagc tagcgatgaa acaaatgaaa 420
 atgagactcc gtattcaaca gatgaaagtg aaaaatcttc ccagtcacat cacaatgaaa 480
 acaactgcac aaaagcaaga ttctctccta aaaacttacg gtttcccatc ccaccaacac 540
 ctccaggatt gggaaggcat ggttcaaaat tcagaacact tccaccattc ttgtcttgct 600
 ggccccacc ctttccagca ggaccaccgt tgattcctcc tccaccacct atggggccag 660
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aagcctatct aaaatag 797

<210> 10
<211> 264
<212> PRT
<213> Gallus gallus

<400> 10

Met	Ala	Gly	Arg	Val	Leu	Phe	Arg	Arg	Gly	Ala	Gly	Gln	Ser	Asp	Asp	1	5	10	15
Ser	Asp	Met	Trp	Asp	Asp	Thr	Ala	Leu	Ile	Lys	Ala	Tyr	Asp	Lys	Ala	20	25	30	
Val	Ala	Ser	Phe	Lys	Asn	Ala	Leu	Lys	Asn	Gly	Asp	Cys	Ser	Glu	Pro	35	40	45	
Ser	Asp	Lys	Gln	Glu	Gln	Arg	Ala	Gly	Val	Lys	Arg	Lys	Asn	Ser	Lys	50	55	60	
Lys	Asn	Arg	Asn	Arg	Asn	Lys	Ser	Asn	Ala	Val	Pro	Leu	Lys	Gln	Trp	65	70	75	80
Lys	Val	Gly	Asp	Ser	Cys	Asn	Ala	Val	Trp	Ser	Glu	Asp	Gly	Asn	Val	85	90	95	
Tyr	Pro	Ala	Thr	Ile	Ala	Ser	Ile	Asn	Leu	Lys	Arg	Gly	Thr	Cys	Val	100	105	110	
Val	Thr	Tyr	Thr	Gly	Tyr	Gly	Asn	Lys	Glu	Glu	Gln	Asn	Leu	Ala	Asp	115	120	125	
Leu	Leu	Pro	Pro	Ala	Ser	Asp	Glu	Thr	Asn	Glu	Asn	Glu	Thr	Pro	Tyr	130	135	140	
Ser	Thr	Asp	Glu	Ser	Glu	Lys	Ser	Ser	Gln	Ser	His	His	Asn	Glu	Asn	145	150	155	160
Asn	Cys	Thr	Lys	Ala	Arg	Phe	Ser	Pro	Lys	Asn	Leu	Arg	Phe	Pro	Ile	165	170	175	
Pro	Pro	Thr	Pro	Pro	Gly	Leu	Gly	Arg	His	Gly	Ser	Lys	Phe	Arg	Thr	180	185	190	
Leu	Pro	Pro	Phe	Leu	Ser	Cys	Trp	Pro	Pro	Pro	Phe	Pro	Ala	Gly	Pro	195	200	205	
Pro	Leu	Ile	Pro	Pro	Pro	Pro	Pro	Met	Gly	Pro	Asp	Ser	Pro	Glu	Asp	210	215	220	
Asp	Glu	Ala	Leu	Gly	Ser	Met	Leu	Ile	Ala	Trp	Tyr	Met	Ser	Gly	Tyr	225	230	235	240

His Thr Gly Tyr Tyr Leu Gly Leu Lys Gln Ser Arg Met Glu Ala Ala
245 250 255

Leu Glu Arg Glu Ala Tyr Leu Lys
260

<210> 11
<211> 407
<212> PRT
<213> Homo sapiens

<400> 11

Met Ser Gly Gly Ser Ala Asp Tyr Asn Arg Glu His Gly Gly Pro Glu
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Gly Met Asp Pro Asp Gly Val Ile Glu Ser Asn Trp Asn Glu Ile Val
20 25 30

Asp Asn Phe Asp Asp Met Asn Leu Lys Glu Ser Leu Leu Arg Gly Ile
35 40 45

Tyr Ala Tyr Gly Phe Glu Lys Pro Ser Ala Ile Gln Gln Arg Ala Ile
50 55 60

Ile Pro Cys Ile Lys Gly Tyr Asp Val Ile Ala Gln Ala Gln Ser Gly
65 70 75 80

Thr Gly Lys Thr Ala Thr Phe Ala Ile Ser Ile Leu Gln Gln Leu Glu
85 90 95

Ile Glu Phe Lys Glu Thr Gln Ala Leu Val Leu Ala Pro Thr Arg Glu
100 105 110

Leu Ala Gln Gln Ile Gln Lys Val Ile Leu Ala Leu Gly Asp Tyr Met
115 120 125

Gly Ala Thr Cys His Ala Cys Ile Gly Gly Thr Asn Val Arg Asn Glu
130 135 140

Met Gln Lys Leu Gln Ala Glu Ala Pro His Ile Val Val Gly Thr Pro
145 150 155 160

Gly Arg Val Phe Asp Met Leu Asn Arg Arg Tyr Leu Ser Pro Lys Trp
165 170 175

Ile Lys Met Phe Val Leu Asp Glu Ala Asp Glu Met Leu Ser Arg Gly
180 185 190

Phe Lys Asp Gln Ile Tyr Glu Ile Phe Gln Lys Leu Asn Thr Ser Ile
195 200 205

Gln Val Val Leu Leu Ser Ala Thr Met Pro Thr Asp Val Leu Glu Val
210 215 220

Thr Lys Lys Phe Met Arg Asp Pro Ile Arg Ile Leu Val Lys Lys Glu
225 230 235 240

Glu Leu Thr Leu Glu Gly Ile Lys Gln Phe Tyr Ile Asn Val Glu Arg
 245 250 255
 Glu Glu Trp Lys Leu Asp Thr Leu Cys Asp Leu Tyr Glu Thr Leu Thr
 260 265 270
 Ile Thr Gln Ala Val Ile Phe Leu Asn Thr Arg Arg Lys Val Asp Trp
 275 280 285
 Leu Thr Glu Lys Met His Ala Arg Asp Phe Thr Val Ser Ala Leu His
 290 295 300
 Gly Asp Met Asp Gln Lys Glu Arg Asp Val Ile Met Arg Glu Phe Arg
 305 310 315 320
 Ser Gly Ser Ser Arg Val Leu Ile Thr Thr Asp Leu Leu Ala Arg Gly
 325 330 335
 Ile Asp Val Gln Gln Val Ser Leu Val Ile Asn Tyr Asp Leu Pro Thr
 340 345 350
 Asn Arg Glu Asn Tyr Ile His Arg Ile Gly Arg Gly Gly Arg Phe Gly
 355 360 365
 Arg Lys Gly Val Ala Ile Asn Phe Val Thr Glu Glu Asp Lys Arg Ile
 370 375 380
 Leu Arg Asp Ile Glu Thr Phe Tyr Asn Thr Thr Val Glu Glu Met Pro
 385 390 395 400
 Met Asn Val Ala Asp Leu Ile
 405